Life with r(20)—Ring chromosome 20 syndrome

Allison Watson, David Watson, and Carol Taylor

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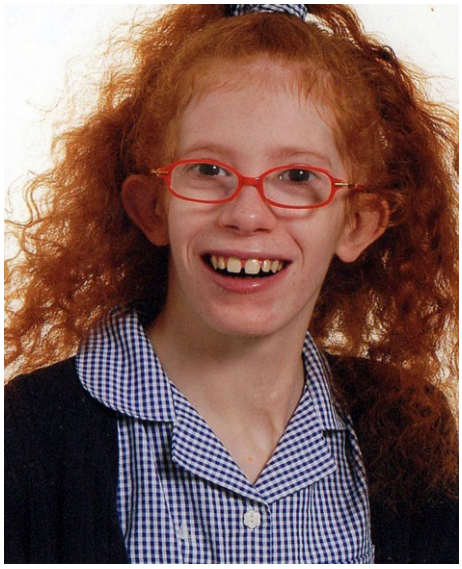
Allison's Story

My 17-year-old son, David, has ring chromosome 20 syndrome—r(20) syndrome—an ultra-rare disease, the main symptom being intractable epilepsy. Rings may be present in all (nonmosaic), or many (mosaic), of the patient's cells. Seizures often start suddenly following normal development and can be severe and frequent. Patients with r(20) syndrome don't respond well to the epilepsy treatments available, and many are on put on multiple antiepileptic drugs (AEDs) and/or recommended for brain surgery workup, for which they're unsuitable. Some patients are much disabled by the condition and/or the multiple medications, rendering them unable to walk or talk. The condition is perceived to be underdiagnosed, and appropriate diagnostic testing is required.

David Watson, aged 17 years, awarded his brown belt in karate.



Rachel Taylor, aged 16 years, going to school.

Allison Watson and Don Gordon at the launch of Ring20 Research and Support UK, at Young Epilepsy, Surrey, United Kingdom, May 2014.

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Ring20 Research and Support UK,Essex,United Kingdom
Address correspondence to Allison Watson, Ring20 Research and Support UK, 26 Headley Chase, Warley, Brentwood, Essex CM14 5BN, U.K. E-mail: ring20uk@outlook.com

David's Story

I had my first seizure at age 6½; after many tests I was told that I had absence seizures. Just before my 7th birthday, I suddenly began to have hourly focal dyscognitive seizures, worse at night, with hallucinating and shouting out. I've tried lots of seizure medications, have a vagus nerve stimulator (VNS), and am about to try the modified ketogenic diet. My seizures initially improved after several months when I was put on a course of steroids, but since my teens, my seizures seem to be longer and more frequent again, lasting 15–20 min, 3–4 per day. Triggers include sports, bathing/showering, eating, tiredness, and stress (e.g., exams). I am fully supported at college by a dedicated learning support assistant, where I am studying engineering despite my seizures. I've attained a brown belt in karate, and last year I spent 2 weeks volunteering in Kenya with the Explorer Scouts. I'm often angry or need a nap after a seizure and am confused when I recover. I experience no warning or aura; seizures occur at any time. As a teen my epilepsy curtails my independence.

Rachel's Story by Carol

The first sign was when I was 22 weeks pregnant and the baby wasn't growing as it should. Rachel was born 2 weeks late and weighed only 5 lb. When she was born I thought something was wrong; she didn't look anything like my other children. As the weeks passed I went to our general practitioner, who examined her and didn't think there was anything wrong but referred her to a consultant who, after blood tests, found that Rachel had r(20) syndrome. She was 6 months old, and at that time there were only 27 reported cases in the world. My nephew is a geneticist in London and he got me some information on the disorder; my consultant told me very little.

Rachel never made the usual milestones. She started special nursery school when she was 3. In her younger years she used to scream a lot and hold her head; looking back I think these episodes were some kind of seizure. She had her first tonic–clonic seizure when she was 18 months old, and then didn't have another until she was 7 when things changed dramatically. Her tonic–clonic seizures would go into focal dyscognitive seizures. They would always start in the early hours of the morning; she would then go on to have behavioral issues that would last all day and night. She would go for 36 to 48 h without sleep. Sometimes she would need midazolam and/or an ambulance—this would happen about every 6 weeks despite Rachel's being on a number of epilepsy drugs. Over the years it went from 6 or 8 weeks down to every 2 weeks. It is now every 6 or 7 days; still the same pattern.

We have tried lots of different combinations of medication. Rachel is currently on four different antiseizure medications. She had a VNS fitted, but the wire got bent so it had to be switched off. She has been on the ketogenic diet, but started choking on her food, so is now waiting for a gastrostomy before going back on the diet. Before Rachel started to have her seizures she used to wake in the night screaming saying "bears," and 4 years ago she was in intensive care unit with her epilepsy; the doctors didn't expect her to live.

It has been very difficult over the years to cope. Rachel is now 17, can just about write her name, has limited speech, and has the ability of a 3- to 4-year-old.

The Charity: "Ring20 Research and Support UK" by Allison

Over the past 10 years, I have made contact with 80+ families around the world with a child with r(20) syndrome, and am in regular contact with many families in the United States who are very communicative, as well as families in the United Kingdom. We share a common need for mutual support and would also welcome more research into the condition. r(20) syndrome has previously been considered a childhood epilepsy syndrome, but as a genetic disorder it stays with the patient for life. Much work is needed to educate adult neurologic teams about the condition, and its impact on quality of life and treatment—many patients are already adults.

I have recently cofounded a new charity with Don Gordon, "Ring20 Research and Support UK," to create a support group forum where families can talk to each other in a secure environment to share experiences and to work together to start to raise funds for much needed research. Our reach extends to families globally. We hope to apply for grants and sponsorship, alongside family fundraising events, and as a patient cohort enable/part-fund any future r(20) research projects (of which we would be interested in any related subject matter).

We are lucky to have input from Dr. Syed Hosain (neurologist) and Nancy Spinner (geneticist), both from the United States, who have previously undertaken research on r(20), from whom we hope to obtain updated information that we can make available on our new website to be launched shortly.

Because our charity operates from within the United Kingdom, we have engaged a UK Health Professional, Dr. Sophia Varadkar, as a main point of contact.

We have created group forums on Facebook and a map of known r(20) cases around the world: https://mapsengine.google.com/map/embed?mid=zg_ssVj-paBI.k30dUsbfkmA4. This allows folks to see the global picture and helps recognize families internationally, who can very often feel isolated and alone.

In May we hosted a UK families meeting, kindly facilitated by Young Epilepsy as an adjunct to their complex epilepsy conference, and were privileged to have a UK

pediatric neurologist and specialist epilepsy nurse attending. I strongly believe that if epilepsy charities can work together, we can learn from and support each other's causes. Recently attending a Findacure workshop, I learned that rare diseases are "fundamental" diseases, in that finding answers to the rare can help the more common. This belief only strengthens our quest.

For more information: https://www.facebook.com/Ring20ResearchandSupportUK

E-mail us: ring20uk@outlook.com

Website: www.ring20researchsupport.co.uk

We are keen to recruit new members, so please pass on our details to any r(20) patients you care for.

Disclosure

The authors declare no conflicts of interest. We confirm that we have read the Journal's position on issues involved in ethical publication and affirm that this report is consistent with those guidelines.

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